ClinVar

NCBI's ClinVar is a freely available submission-driven database for information about genomic variation and its relationship to human health.

ClinVar accepts submissions interpretations of genetic data from:

- **clinical genetics testing laboratories**
- **research groups**
- **expert panels**
- **and others**

Interpret your data and guide your diagnosis

ncbi.nlm.nih.gov/clinvar

- **1,670+ submitters**
- **75+ countries**
- **841,000+ variants**
- **1,300,000+ submitted records**
- **ClinVar Search Video**

Contact us at clinvar@ncbi.nlm.nih.gov

Follow us on Twitter @ncbi_clinical

Visit us at ncbi.nlm.nih.gov/clinvar/ to find out more

National Library of Medicine
National Center for Biotechnology Information

ClinVar aggregates

clinical assertions about variants provided by clinical genetics testing laboratories

and others.

ClinVar helps clinicians

interpret genetic test results

and diagnose disorders to

improve patient outcomes.

What's New

Automated validation in the ClinVar Submission Portal for quick resolution of common errors and faster submission processing time

Tailored notifications for changes in clinical interpretation of variants

Submit to ClinVar

Submit Now

1. **Setup and register** – Go to ClinVar Submission Portal to create your myncbi account and register your organization
2. **Submit** – Use the submission wizard for a single variant submission or excel, TSV/CSV, or XML formats for multiple submissions
3. **Review and access** –.Your data will be available on ClinVar after curatorial review and processing

Dowload data from the ClinVar FTP Site

dbGaP

An NIH-sponsored repository for archiving, curating, and distributing information produced by genome-scale studies investigating the interaction of human genotype and phenotype

Augment your research

View Map

Over
2.6 million
research subjects

Over
1,500 research studies

Over
350,000 variables

Over
100,000
samples of non-genomics omics data

Over
400,000
whole genome and whole exome sequences related to dbGaP studies, available on Amazon Web Services and Google Cloud

dbGaP study submission steps (NIH funded studies)

1 Registration

- Contact NIH Program Officer or Genomic Program Administrator (GPA)
- Receive invitation
- Enter study metadata

2 Submission

- Use dbGaP submission guide to upload files
- Work with curators to complete submission
- Get accession number

3 Release

- Approve processed data
- Release study

Submit Now

Upcoming

- Public API for study metadata and controlled-access data access using FHIR (Fast Healthcare Interoperability Resources) protocol
- Automated validation in dbGaP Submission Portal for quick feedback and shorter submission processing timeframes

dbGaP study access steps (for Principal Investigators (PIs)

1 Account Setup

- NIH Intramural researchers – submit permission form to establish data request eligibility in dbGaP
- Other researchers – Get eRA commons user account

2 Access Application

- Complete / revise and submit application to Signing Officer (SO)
- SO certifies application with one or more Data Access Requests (DAR)

3 Approval and Access

- dbGaP Data Access Committee (DAC) reviews and approves application
- dbGaP approved data is provided for download

NIH Genetic Testing Registry (GTR®)

An unbiased, free to participate in, and free to use, international database of clinical and research molecular, cytogenetics and biochemical genetic tests, and supporting information

ABOUT GTR®

- Single gene tests, panels, genomes, and exomes
- 76,000+ tests (incl. 1,600+ tests for somatic targets), 16,000+ conditions, 18,500+ genes, 575+ labs
- 56 BRCA1 single gene tests and 391 multigene panels
- 78 BRCA2 single gene tests and 445 multigene panels

GTR® now includes molecular and serological tests for microbes that affect human health & disease

- 18 COVID-19 tests
- 15 tests for other viruses, parasites, bacteria

GTR® is a central location for laboratories to provide genetic test information and for clinicians and researchers to search and find genetic tests. GTR® increases transparency in the genetic testing landscape.

Search by

- Test name
- Test services like custom mutation-specific / carrier testing
- Gene, number of genes, or germline vs. somatic
- Analytes / chromosomal regions / proteins
- Lab and staff name, location, or certifications
- Test purpose or specimen type
- Disease or phenotype
- Methodologies

Starting October 2020, register your microbe tests, including:

- molecular tests to detect microbe nucleic acids
- tests to detect microbe-specific antigens
- tests to detect antibodies to a microbe
- microbe panels
- viral load tests to monitor disease progression and guide treatment

LEARN ABOUT GENETIC TESTS AVAILABLE TO YOU

Visit GTR

- Purpose and limitations
- Clinical utility
- Methodology
- Clinical and analytical validity
- Lab contacts and credentials, including CLIA and state licenses
- AMA CPT® and LOINC codes
- Evidence of the test's usefulness
- Test ordering information

MedGen

Medical genetics information at your fingertips, aggregated from your favorite resources!

ACCESS RESOURCES

Gain centralized access to genetic disease, phenotype data, and analytical tools from authoritative resources to learn about the clinically actionable information currently available in your area of interest

SEARCH CLINICAL FEATURES

Search for genetic phenotype information by clinical features, genes or other attributes. Access aggregated phenotype data from HPO, MONDO, OMIM, UMLS, and others, and use their identifiers harmonized for you

INFORM YOUR RESEARCH

Access the latest research in your area of interest, including the latest clinical studies, systematic reviews, and practice guidelines from medical and professional societies that provide effective treatment options for patients

RESOURCES

MedGen supports research, diagnosis and treatment of genetic disorders by providing information on:

- Mendelian disorders
- Pharmacogenetic responses
- Complex diseases
- Clinical findings

TOOLS

MedGen's all-in-one platform connects clinicians to leading genetic resources, including:

- PubMed
- GARD
- GeneReviews®
- OMIM

Visit MedGen

National Library of Medicine
National Center for Biotechnology Information

Follow us on Twitter
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Contact us at
info@ncbi.nlm.nih.gov

Visit us at
ncbi.nlm.nih.gov/medgen/

MedGen, ClinVar, and GTR®

Using NCBI's medical genetics and human variation resources to research, diagnose and treat genetic conditions.

CASE STUDY: SUSPECTED MARFAN SYNDROME

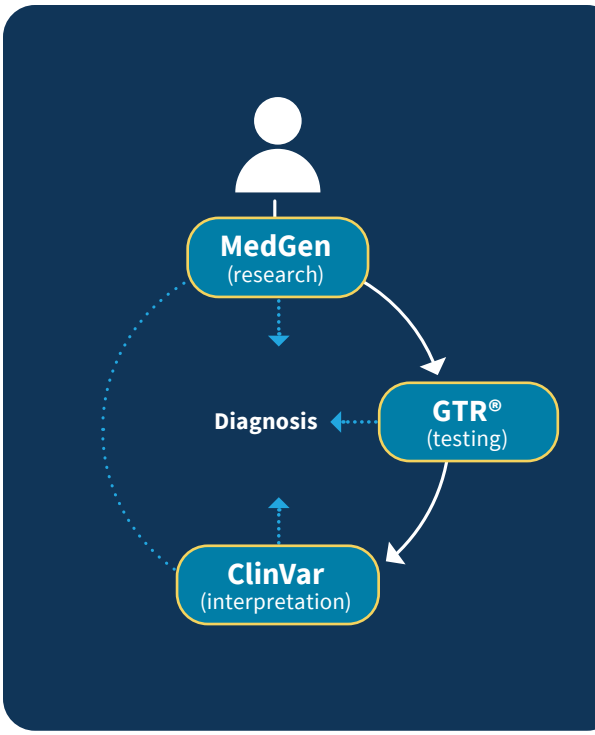
A 9-year-old boy comes into the clinic for a medical release to play soccer. The child presents with myopia, arachnodactyly and has a family history of aortic dissection. These are flags that prompt you to investigate a possible genetic disease.

1 Search MedGen to help develop a differentby patient's clinical features in ial diagnosis and learn about a condition, its diagnosis and etiology

2 Search the NIH Genetic Testing Registry (GTR®) to find the most appropriate genetic test for your patient – for example a panel that includes all the conditions in your differential diagnosis. Learn about the test's validity and utility, and find the ordering information

3 Following testing, compare the test results to interpreted variants in ClinVar and determine pathogenicity

4 Return to MedGen for practice guidelines from medical and professional societies and the latest research to guide your treatment options. Access consumer resources to help your patient and his family understand his diagnosis, prognosis and available help. If the family is interested, check if there are available studies in ClinicalTrials.gov



ABOUT OUR RESOURCES

ClinVar has more than 1,300,000 submitted records representing more than 841,000 unique variants from 1670+ submitters.

GTR® has 76,000+ tests for 16,000+ conditions and 18,500+ genes, from 575+ labs. It includes clinical and research molecular, cytogenetic and biochemical genetic tests.

MedGen helps research thousands of genetic phenotypes including Mendelian disorders, complex diseases, clinical features and drug responses. It aggregates information from authoritative resources so from one website you can access most available clinical, consumer and molecular resources.

Share Your Data with ClinVar

Share Your Data with GTR

Variation Resources

NCBI's variation resources offer human genomic variations, including common and rare SNV, other small-scale variations, large structural variations, and associated frequencies, including ALFA, a new aggregated frequency source based on data from millions of controlled-access research study subjects. Access through the web, APIs, and FTP downloads.

Identify novel variants

Annotate with other data such as genomic features, Genes & Pubmed citations

Integrate into analysis tools and workflows

Prioritize variants likely to impact biological function

dbSNP

Visit dbSNP

- Over 2 Billion submissions including data from 1000 Genomes, GnomAD, and others
- 720 Million RS
- Frequency for more than 606 Million RS; including common and rare variants
- Rich annotation reported on RefSeq GRCH37 and GRCH38 assemblies, mRNA, and Protein
- VCF files for assemblies GRCh37 and GRCh38
- Full set of RefSNPs in the JSON format
- Indexed Search

dbVar

Visit dbVar

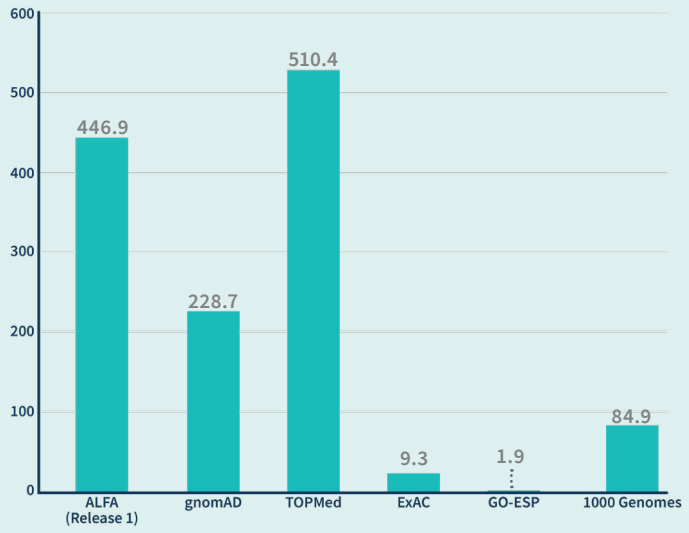
- 193 studies
- Clinically significant SV, Case-Control, and Curated Datasets
- 6.0 million unique structural variants
- 36.1 million submitted variant calls
- Population allele frequency
- Updated monthly
- Files are available in XML, GVF, VCF, BED, BEDPE, and TSV for assemblies GRCh37 and GRCh38
- dbVar Tutorials and Datasets
- Access full set of FTP files

ALFA

Visit ALFA

- Release 1 (March 2020) included 447M variants from 98K subjects
- Release 2 (October 2020) will include an additional ~100K subjects for a total of ~200K
- Access ALFA data along with other projects including GnomAD, and TOPMed

Variants with frequency data (by project in, million)



Variation Services

Web services for comparing, normalizing, annotating, and inter-converting variations

Visit Now

Variation Viewer

View, search, and navigate variations in genomic context. Review data or upload your own data

Visit Now

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Visit us at ncbi.nlm.nih.gov/variation/